Format: Abstract

Hum Genome Var. 2019 Apr 26;6:21. doi: 10.1038/s41439-019-0052-z. eCollection 2019.

Two closely spaced mutations *in cis* result in Ullrich congenital muscular dystrophy.

Shimomura H[1], Lee T[1], Tanaka Y[1], Awano H[2], Itoh K[3], Nishino I[4], Takeshima Y[1].

Author information

Abstract

A 2-year-old boy was diagnosed with Ullrich congenital muscular dystrophy (UCMD) by muscle biopsy. COL6A3 gene analysis by next-generation sequencing revealed two heterozygous splice-site mutations (c.6283-1 G > G/T and c.6310-2 A > A/T), whereas normal mRNA was produced. Genomic DNA analysis revealed two mutations located on the same allele; however, no mutation was detected in either parent. These results indicated that two closely spaced de novo mutations resulted in the autosomal dominant UCMD.

KEYWORDS: Disease genetics; Neuromuscular disease

PMID: 31044083 PMCID: PMC6486579 DOI: 10.1038/s41439-019-0052-z

Conflict of interest statement